

Human chromosomal abnormalities

Chromosomes are tightly wound-up coils of DNA and are found in every cell in our bodies. When a cell divides, all of the chromosomes are copied and split between the two cells, but sometimes things can go wrong with this copying process. For example, cells can end up with too many or too few chromosomes. These are called chromosomal abnormalities.

An organised profile of someone's chromosomes is called a karyotype. Karyotyping is one of many techniques that can detect chromosomal abnormalities, by looking at the number and structure of chromosomes in cells.

In this activity, you will make your own karyotypes and look at the patterns for three chromosomal abnormalities.

You will need:

- Read *The full set* from *What is Biotechnology?* in *Biotechnology Online*
- Scissors, glue and paper
- The two master ("normal") karyotypes and three random chromosome patterns supplied on the following pages

What to do:

1. Working in pairs, or groups of three, cut out each chromosome from the pages with random chromosome patterns using a pair of scissors.
2. Using the male and female master karyotypes for comparison, sort and match the chromosomes together to produce karyotypes on a new piece of paper for a:
 - male with Edward Syndrome (trisomy 18)
 - female with Down syndrome (trisomy 21)
 - male with Klinefelter syndrome (XXY)

How do each of the karyotypes differ?

For further investigation

Do some research into two other chromosomal abnormalities:

- what is the chromosomal change or abnormality?
- how are people with that abnormality affected?
- are any of these diseases treatable?

Think about why chromosomal abnormalities happen – what physical processes might cause there to be an abnormal number of chromosomes?

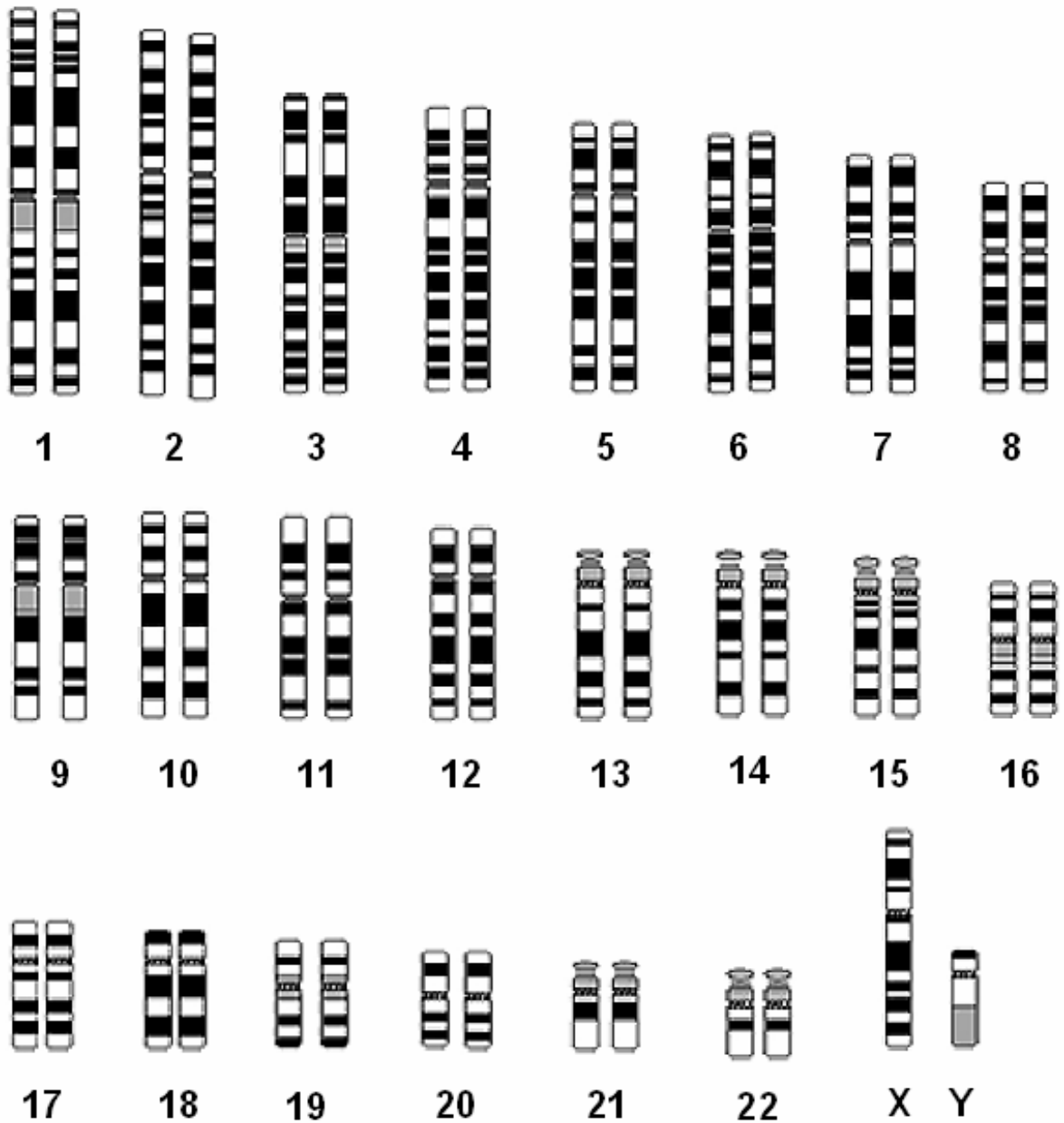
Are there any technologies available to fix these abnormalities, and what are they?



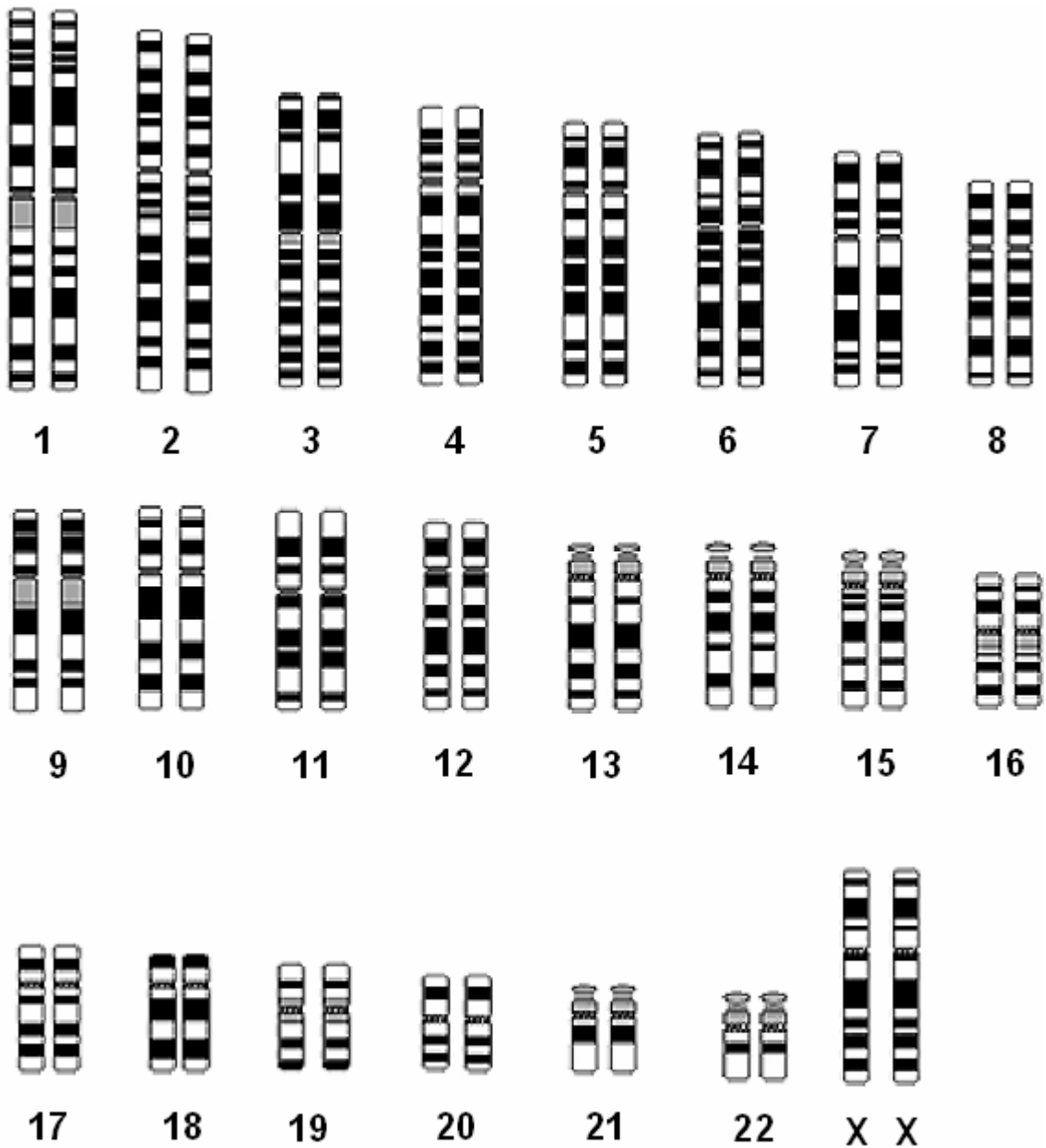
This work sheet was adapted from the Children's Medical Research institute.

For more information on the CMRI's education programs go to www.cmri.com.au.

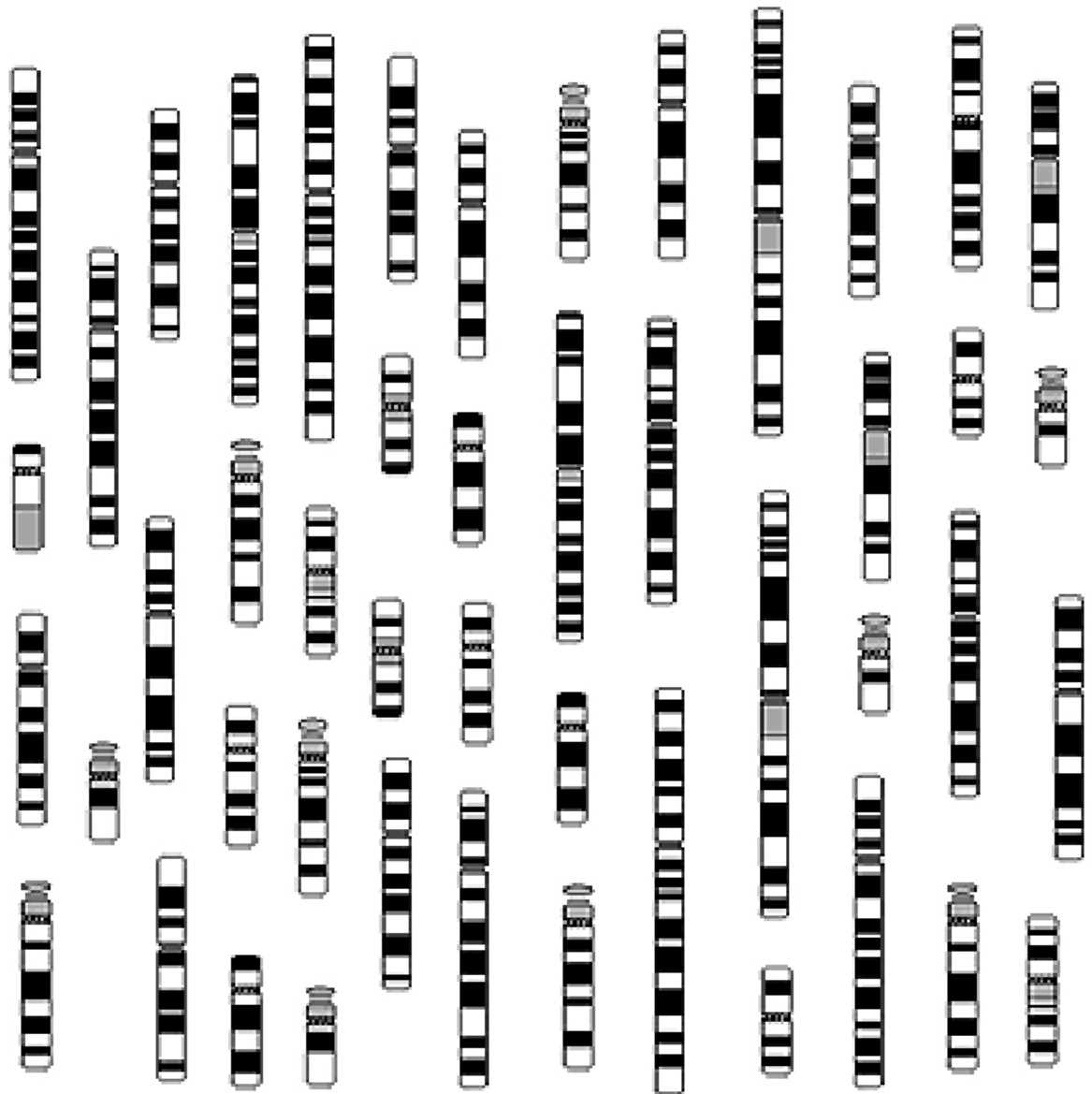
Male karyotype - master



Female karyotype - master



Male with Edward syndrome (trisomy 18)

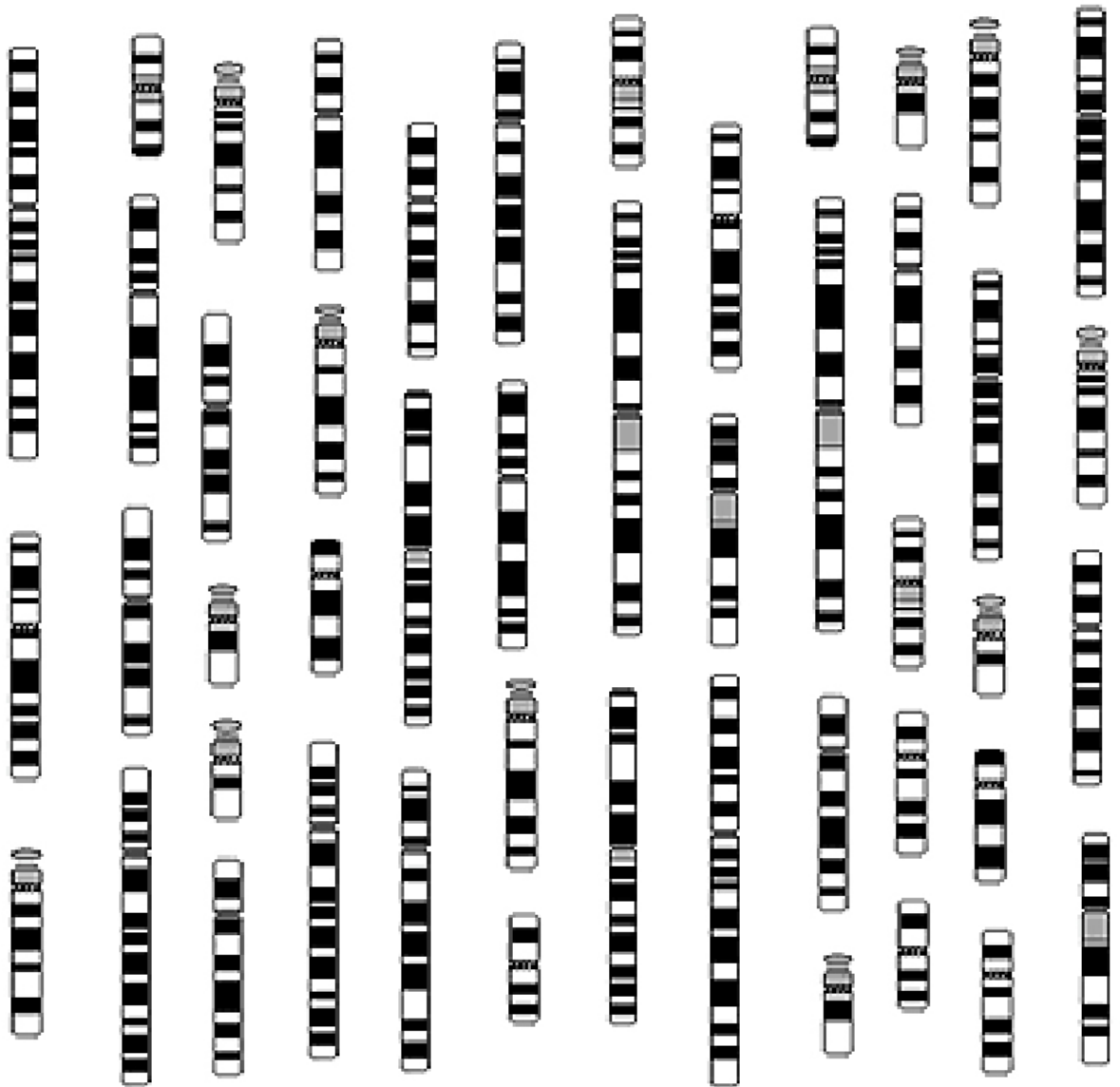


Edward syndrome

Incidence: 1:5000 live births, 1:50,000 stillbirths

A defect present at birth caused by an extra copy of chromosome 18 (trisomy 18). Characteristics of the disorder include a number of malformed organs, including heart defects, malformed features of the face and skeleton and motor and mental retardation. In most cases, the child dies before it is born. 90% of babies born alive die within a year of birth.

Female with Down syndrome (trisomy 21)



Down Syndrome

Incidence: 1 in 660 births.

A defect present at birth, in which a person is born with three copies of chromosome 21 (trisomy 21).

Clinical features include moderate to severe mental retardation, slanting eyes, a broad short skull, broad hands and short fingers. Other abnormalities include heart defects, oesophageal atresia (oesophagus not connected with the stomach) and an increased chance of developing acute lymphocytic leukaemia.

Male with Klinefelter syndrome (XXY)



Klinefelter syndrome

Incidence: 1 in 500 to 1 in 1000 newborn males.

Klinefelter only affects males and is caused by an extra X chromosome. Males normally have one X chromosome and one Y, but the Klinefelter karyotype is XXY (or more rarely XXXY, XXXXY or sometimes XY/XXY mosaic). The child appears normal at birth, but the syndrome usually becomes apparent during puberty. As Klinefelter syndrome males produce small amounts of testosterone and high amounts of oestrogen, they usually have high-pitched voices, can look more like a woman than a man, have relatively little facial and body hair and are infertile. However, it is not unusual for Klinefelter syndrome adults with slight symptoms to be unaware that they have it until they are tested for infertility. Klinefelter syndrome males with more than two X chromosomes usually have more extreme symptoms and are often mentally retarded.